

SEQUENCE LISTING

<110> Dean, Michael
Allikmets, Rando
Hutchinson, Amy A.

<120> ATP-BINDING TRANSPORTER (ABC7) AND METHODS FOR DETECTION OF ANEMIA AND ATAXIA

<130> 4239-67289

<150> US 09/422,840
<151> 1999-10-21

<150> US 60/105,497
<151> 1998-10-23

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<170> PatentIn version 3.1

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acatt	2345

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 <212> PRT
 <213> Homo sapien

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Gly Pro Gln Trp Arg Pro His Gln Leu Gly Ala Leu Gly Thr Ala Arg
35 40 45

Ala Tyr Gln Ile Pro Glu Ser Leu Lys Ser Ile Thr Trp Gln Arg Leu
50 55 60

Gly Lys Gly Asn Ser Gly Gln Phe Leu Asp Ala Ala Lys Ala Leu Gln
65 70 75 80

Val Trp Pro Leu Ile Glu Lys Arg Thr Cys Trp His Gly His Ala Gly
85 90 95

Gly Gly Leu His Thr Asp Pro Lys Glu Gly Leu Lys Asp Val Asp Thr
100 105 110

Arg Lys Ile Ile Lys Ala Met Leu Ser Tyr Val Trp Pro Lys Asp Arg
115 120 125

Pro Asp Leu Arg Ala Arg Val Ala Ile Ser Leu Gly Phe Leu Gly Gly
130 135 140

Ala Lys Ala Met Asn Ile Val Val Pro Phe Met Phe Lys Tyr Ala Val
145 150 155 160

Asp Ser Leu Asn Gln Met Ser Gly Asn Met Leu Asn Leu Ser Asp Ala
165 170 175

Pro Asn Thr Val Ala Thr Met Ala Thr Ala Val Leu Ile Gly Tyr Gly
180 185 190

Val Ser Arg Ala Gly Ala Ala Phe Phe Asn Glu Val Arg Asn Ala Val
195 200 205

Phe Gly Lys Val Ala Gln Asn Ser Ile Arg Arg Ile Ala Lys Asn Val
210 215 220

Phe Leu His Leu His Asn Leu Asp Leu Gly Phe His Leu Ser Arg Gln
225 230 235 240

Thr Gly Ala Leu Ser Lys Ala Ile Asp Arg Gly Thr Lys Gly Ile Ser
245 250 255

Phe Val Leu Ser Ala Leu Val Phe Asn Leu Leu Pro Ile Met Phe Glu
260 265 270

Val Met Leu Val Ser Gly Val Leu Tyr Tyr Lys Cys Gly Ala Gln Phe
275 280 285

Ala Leu Val Thr Leu Gly Thr Leu Gly Thr Tyr Thr Ala Phe Thr Val
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Ala Val Thr Arg Trp Arg Thr Arg Phe Arg Ile Glu Met Asn Lys Ala
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Asp Asn Asp Ala Gly Asn Ala Ala Ile Asp Ser Leu Leu Asn Tyr Glu
325 330 335

Thr Val Lys Tyr Phe Asn Asn Glu Arg Tyr Glu Ala Gln Arg Tyr Asp
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Gly Phe Leu Lys Thr Tyr Glu Thr Ala Ser Leu Lys Ser Thr Ser Thr
355 360 365

Leu Ala Met Leu Asn Phe Gly Gln Ser Ala Ile Phe Ser Val Gly Leu
370 375 380

Thr Ala Ile Met Val Leu Ala Ser Gln Gly Ile Val Ala Gly Thr Leu
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Thr Val Gly Asp Leu Val Met Val Asn Gly Leu Leu Phe Gln Leu Ser
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Leu Pro Leu Asn Phe Leu Gly Thr Val Tyr Arg Glu Thr Arg Gln Ala
420 425 430

Leu Ile Asp Met Asn Thr Leu Phe Thr Leu Leu Lys Val Asp Thr Gln
435 440 445

Ile Lys Asp Lys Val Met Ala Ser Pro Leu Gln Ile Thr Pro Gln Thr

450 455 460

Ala Thr Val Ala Phe Asp Asn Val His Phe Glu Tyr Ile Glu Gly Gln
465 470 475 480

Lys Val Leu Ser Gly Ile Ser Phe Glu Val Pro Ala Gly Lys Lys Val
485 490 495

Ala Ile Val Gly Gly Ser Gly Ser Gly Lys Ser Thr Ile Val Arg Leu
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Leu Phe Arg Phe Tyr Glu Pro Gln Lys Gly Ser Ile Tyr Leu Ala Gly
515 520 525

Gln Asn Ile Gln Asp Val Ser Leu Glu Ser Leu Arg Arg Ala Val Gly
530 535 540

Val Val Pro Gln Asp Ala Val Leu Phe His Asn Thr Ile Tyr Tyr Asn
545 550 555 560

Leu Leu Tyr Gly Asn Ile Ser Ala Ser Pro Glu Glu Val Tyr Ala Val
565 570 575

Ala Lys Leu Ala Gly Leu His Asp Ala Ile Leu Arg Met Pro His Gly
580 585 590

Tyr Asp Thr Gln Val Gly Glu Arg Gly Leu Lys Leu Ser Gly Gly Glu
595 600 605

Lys Gln Arg Val Ala Ile Ala Arg Ala Ile Leu Lys Asp Pro Pro Val
610 615 620

Ile Leu Tyr Asp Glu Ala Thr Ser Ser Leu Asp Ser Ile Thr Glu Glu
625 630 635 640

Thr Ile Leu Gly Ala Met Lys Asp Val Val Lys His Arg Thr Ser Ile
645 650 655

Phe Ile Ala His Arg Leu Ser Thr Val Val Asp Ala Asp Glu Ile Ile
660 665 670

Val Leu Asp Gln Gly Lys Val Ala Glu Arg Gly Thr His His Gly Leu
675 680 685

Leu Ala Asn Pro His Ser Ile Tyr Ser Glu Met Trp His Thr Gln Ser
690 695 700

Ser Arg Val Gln Asn His Asp Asn Pro Lys Trp Glu Ala Lys Lys Glu
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725 730 735

Asn Ser Val Lys Gly Cys Gly Asn Cys Ser Cys
740 745

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Gly Pro Gln Trp Arg Pro His Gln Leu Gly Ala Leu Gly Thr Ala Arg
35 40 45

Ala Tyr Gln Ile Pro Glu Ser Leu Lys Ser Ile Thr Trp Gln Arg Leu
50 55 60

Gly Lys Gly Asn Ser Gly Gln Phe Leu Asp Ala Ala Lys Ala Leu Gln
65 70 75 80

Val Trp Pro Leu Ile Glu Lys Arg Thr Cys Trp His Gly His Ala Gly
85 90 95

Gly Gly Leu His Thr Asp Pro Lys Glu Gly Leu Lys Asp Val Asp Thr
100 105 110

Arg Lys Ile Ile Lys Ala Met Leu Ser Tyr Val Trp Pro Lys Asp Arg
115 120 125

Pro Asp Leu Arg Ala Arg Val Ala Ile Ser Leu Gly Phe Leu Gly Gly
130 135 140

Ala Lys Ala Met Asn Ile Val Val Pro Phe Met Phe Lys Tyr Ala Val
145 150 155 160

Asp Ser Leu Asn Gln Met Ser Gly Asn Met Leu Asn Leu Ser Asp Ala
165 170 175

Pro Asn Thr Val Ala Thr Met Ala Thr Ala Val Leu Ile Gly Tyr Gly
180 185 190

Val Ser Arg Ala Gly Ala Ala Phe Phe Asn Glu Val Arg Asn Ala Val
195 200 205

Phe Gly Lys Val Ala Gln Asn Ser Ile Arg Arg Ile Ala Lys Asn Val
210 215 220

Phe Leu His Leu His Asn Leu Asp Leu Gly Phe His Leu Ser Arg Gln
225 230 235 240

Thr Gly Ala Leu Ser Lys Ala Ile Asp Arg Gly Thr Lys Gly Ile Ser
245 250 255

Phe Val Leu Ser Ala Leu Val Phe Asn Leu Leu Pro Ile Met Phe Glu
260 265 270

Val Met Leu Val Ser Gly Val Leu Tyr Tyr Lys Cys Gly Ala Gln Phe
275 280 285

Ala Leu Val Thr Leu Gly Thr Leu Gly Thr Tyr Thr Ala Phe Thr Val
290 295 300

Ala Val Thr Arg Trp Arg Thr Arg Phe Arg Ile Glu Met Asn Lys Ala
305 310 315 320

Asp Asn Asp Ala Gly Asn Ala Ala Ile Asp Ser Leu Leu Asn Tyr Glu
325 330 335

Thr Val Lys Tyr Phe Asn Asn Glu Arg Tyr Glu Ala Gln Arg Tyr Asp
340 345 350

Gly Phe Leu Lys Thr Tyr Glu Thr Ala Ser Leu Lys Ser Thr Ser Thr
355 360 365

Leu Ala Met Leu Asn Phe Gly Gln Ser Ala Ile Phe Ser Val Gly Leu
370 375 380

Thr Ala Ile Met Val Leu Ala Ser Gln Gly Met Val Ala Gly Thr Leu
385 390 395 400

Thr Val Gly Asp Leu Val Met Val Asn Gly Leu Leu Phe Gln Leu Ser
405 410 415

Leu Pro Leu Asn Phe Leu Gly Thr Val Tyr Arg Glu Thr Arg Gln Ala
420 425 430

Leu Ile Asp Met Asn Thr Leu Phe Thr Leu Leu Lys Val Asp Thr Gln
435 440 445

Ile Lys Asp Lys Val Met Ala Ser Pro Leu Gln Ile Thr Pro Gln Thr
450 455 460

Ala Thr Val Ala Phe Asp Asn Val His Phe Glu Tyr Ile Glu Gly Gln
465 470 475 480

Lys Val Leu Ser Gly Ile Ser Phe Glu Val Pro Ala Gly Lys Lys Val
485 490 495

Ala Ile Val Gly Gly Ser Gly Ser Gly Lys Ser Thr Ile Val Arg Leu
500 505 510

Leu Phe Arg Phe Tyr Glu Pro Gln Lys Gly Ser Ile Tyr Leu Ala Gly
515 520 525

Gln Asn Ile Gln Asp Val Ser Leu Glu Ser Leu Arg Arg Ala Val Gly
530 535 540

Val Val Pro Gln Asp Ala Val Leu Phe His Asn Thr Ile Tyr Tyr Asn
545 550 555 560

Leu Leu Tyr Gly Asn Ile Ser Ala Ser Pro Glu Glu Val Tyr Ala Val
565 570 575

Ala Lys Leu Ala Gly Leu His Asp Ala Ile Leu Arg Met Pro His Gly
580 585 590

Tyr Asp Thr Gln Val Gly Glu Arg Gly Leu Lys Leu Ser Gly Gly Glu
595 600 605

Lys Gln Arg Val Ala Ile Ala Arg Ala Ile Leu Lys Asp Pro Pro Val
610 615 620

Ile Leu Tyr Asp Glu Ala Thr Ser Ser Leu Asp Ser Ile Thr Glu Glu
625 630 635 640

Thr Ile Leu Gly Ala Met Lys Asp Val Val Lys His Arg Thr Ser Ile
645 650 655

Phe Ile Ala His Arg Leu Ser Thr Val Val Asp Ala Asp Glu Ile Ile
660 665 670

Val Leu Asp Gln Gly Lys Val Ala Glu Arg Gly Thr His His Gly Leu
675 680 685

Leu Ala Asn Pro His Ser Ile Tyr Ser Glu Met Trp His Thr Gln Ser

690

695

700

Ser Arg Val Gln Asn His Asp Asn Pro Lys Trp Glu Ala Lys Lys Glu
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Asn Ser Val Lys Gly Cys Gly Asn Cys Ser Cys
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Val Ser Lys Leu Ser Thr Gln Arg Pro Leu Leu Phe Asn Ser Ala Val
35 40 45

Asn Leu Trp Asn Gln Ala Gln Lys Asp Ile Thr His Lys Lys Ser Val
50 55 60

Glu Gln Phe Ser Ser Ala Pro Lys Val Lys Thr Gln Val Lys Lys Thr
65 70 75 80

Ser Lys Ala Pro Thr Leu Ser Glu Leu Lys Ile Leu Lys Asp Leu Phe
85 90 95

Arg Tyr Ile Trp Pro Lys Gly Asn Asn Lys Val Arg Ile Arg Val Leu
100 105 110

Ile Ala Leu Gly Leu Leu Ile Ser Ala Lys Ile Leu Asn Val Gln Val
115 120 125

Pro Phe Phe Phe Lys Gln Thr Ile Asp Ser Met Asn Ile Ala Trp Asp
130 135 140

Asp Pro Thr Val Ala Leu Pro Ala Ala Ile Gly Leu Thr Ile Leu Cys
145 150 155 160

Tyr Gly Val Ala Arg Phe Gly Ser Val Leu Phe Gly Glu Leu Arg Asn
165 170 175

Ala Val Phe Ala Lys Val Ala Gln Asn Ala Ile Arg Thr Val Ser Leu
180 185 190

Gln Thr Phe Gln His Leu Met Lys Leu Asp Leu Gly Trp His Leu Ser
195 200 205

Arg Gln Thr Gly Gly Leu Thr Arg Ala Met Asp Arg Gly Thr Lys Gly
210 215 220

Ile Ser Gln Val Leu Thr Ala Met Val Phe His Ile Ile Pro Ile Ser
225 230 235 240

Phe Glu Ile Ser Val Val Cys Gly Ile Leu Thr Tyr Gln Phe Gly Ala
245 250 255

Ser Phe Ala Ala Ile Thr Phe Ser Thr Met Leu Leu Tyr Ser Ile Phe
260 265 270

Thr Ile Lys Thr Thr Ala Trp Arg Thr His Phe Arg Arg Asp Ala Asn
275 280 285

Lys Ala Asp Asn Lys Ala Ala Ser Val Ala Leu Asp Ser Leu Ile Asn
290 295 300

Phe Glu Ala Val Lys Tyr Phe Asn Asn Glu Lys Tyr Leu Ala Asp Lys
305 310 315 320

Tyr Asn Gly Ser Leu Met Asn Tyr Arg Asp Ser Gln Ile Lys Val Ser
325 330 335

Gln Ser Leu Ala Phe Leu Asn Ser Gly Gln Asn Leu Ile Phe Thr Thr
340 345 350

Ala Leu Thr Ala Met Met Tyr Met Gly Cys Thr Gly Val Ile Gly Gly
355 360 365

Asn Leu Thr Val Gly Asp Leu Val Leu Ile Asn Gln Leu Val Phe Gln
370 375 380

Leu Ser Val Pro Leu Asn Phe Leu Gly Ser Val Tyr Arg Asp Leu Lys
385 390 395 400

Gln Ser Leu Ile Asp Met Glu Thr Leu Phe Lys Leu Arg Lys Asn Glu
405 410 415

Val Lys Ile Lys Asn Ala Glu Arg Pro Leu Met Leu Pro Glu Asn Val
420 425 430

Pro Tyr Asp Ile Thr Phe Glu Asn Val Thr Phe Gly Tyr His Pro Asp
435 440 445

Arg Lys Ile Leu Lys Asn Ala Ser Phe Thr Ile Pro Ala Gly Trp Lys
450 455 460

Thr Ala Ile Val Gly Ser Ser Gly Ser Gly Lys Ser Thr Ile Leu Lys
465 470 475 480

Leu Val Phe Arg Phe Tyr Asp Pro Glu Ser Gly Arg Ile Leu Ile Asn
485 490 495

Gly Arg Asp Ile Lys Glu Tyr Asp Ile Asp Ala Leu Arg Lys Val Ile
500 505 510

Gly Val Val Pro Gln Asp Thr Pro Leu Phe Asn Asp Thr Ile Trp Glu
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Asn Val Lys Phe Gly Arg Ile Asp Ala Thr Asp Glu Glu Val Ile Thr
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Val Val Glu Lys Ala Gln Leu Ala Pro Leu Ile Lys Lys Leu Pro Gln

545

550

555

560

Gly Phe Asp Thr Ile Val Gly Glu Arg Gly Leu Met Ile Ser Gly Gly
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Glu Lys Gln Arg Leu Ala Ile Ala Arg Val Leu Leu Lys Asn Ala Arg
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Ile Met Phe Phe Asp Glu Ala Thr Ser Ala Leu Asp Thr His Thr Glu
595 600 605

Gln Ala Leu Leu Arg Thr Ile Arg Asp Asn Phe Thr Ser Gly Ser Arg
610 615 620

Thr Ser Val Tyr Ile Ala His Arg Leu Arg Thr Ile Ala Asp Ala Asp
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Lys Ile Ile Val Leu Asp Asn Gly Arg Val Arg Glu Glu Gly Lys His
645 650 655

Leu Glu Leu Leu Ala Met Pro Gly Ser Leu Tyr Arg Glu Leu Trp Thr
660 665 670

Ile Gln Glu Asp Leu Asp His Leu Glu Asn Glu Leu Lys Asp Gln Gln
675 680 685

Glu Leu
690